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## P 365

**TITLE: IMMUNOCYTOCHEMICAL AND ELECTRON MICROSCOPIC STUDY OF HUMAN PRERETINAL MEMBRANES IN THE PROLIFERATIVE VITREORETINOPATHY.**

AUTHORS: Cano-Parra J[1], Duch A[2], Iborra FJ[2], Ruiz-Lapuente C[1], Martinez-Palmer A[1], Navea A[2], Diaz M[2].
1.- IMIM, Barcelona.
2.- La Fe Hospital, Valencia.

**PURPOSE.** Light microscopic and ultrastructural studies were perfomed in the human epiretinal membranes secondary to proliferative vitreoretinopathy (PVR). **METHODS.** Human preretinal membranes from 5 eyes with PVR were obtained by vitrectomy. These membranes were processed for inmunohistochemical (GFAP) and electron microscopy studies to study the morphology and the cell composition. **RESULTS.** Light microscopic studies manifested a preretinal membrane with indiferenciated glial cells and some macrophage and retinal pigment epithelial cells. Ultrastructural studies showed mainly glial cells with intermediate filaments that stained with GPAP and retinal pigment epithelial cells. The extracellular matrix was mainly composed of collagen.
**CONCLUSIONS.** The glia is the main cell component found in the human preretinal membranes from eyes with postsurgical PVR.

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**PROLIFERATIVE VITREORETINOPATHY (PVR). ULTRASTRUCTURAL AND IMMUNOHISTOCHEMICAL STUDY OF 46 MEMBRANES REMOVED BY VITREOUS SURGERY.**

**PALMIERI G.*, SANTEUSANIO G.*, BLASI M.****
*** Cattedra di Anatomia Patologica, Universita' di Tor Vergata, Roma**
**** Cattedra di Clinica Oculistica, Universita' dell'Aquila, L'Aquila**

***Purpose***
The histopathological peculiarity of PVR is the formation of epi- and subretinal membranes.There are two partially unsolved problems in the histogenesis of PVR. The first concerns the origin and identity of the cellular component and the second problem centers on the individualization of cellular contractile elements, considered in literature retinal pigment epithelium cells with myoblastic differentiation. To characterize the membrane cell populations, we used ultrastructural criteria and immunohistochemical cell markers.
***Methods***
Fourty-six epi- and subretinal membranes were surgically excised from 39 patients (29 males and ten females) affected by PVR, immediately fixed in Karnovsky solution containing 4.5% saccharose and used for ultrastructural and immunohistochemical study.
***Results***
In all the membranes, we observed non metaplastic retinal pigment epithelium, fibroblasts, macrophages, rare glial cells, endothelial cells and myofibroblasts associated with an intercellular matrix consisting of collagen fibrils 100 to 350 nm in diameter and abundant fibrin.
***Conclusions***
Our data indicate that the extracellular matrix of the membranes is synthetized by fibroblasts and myofibroblasts of haematogenous origin and not by metaplastic epithelial cells.. We observed frequently in the membranes endothelial cells arranged to form capillary lumen. The process that determine the membrane formation seems therefore similar to that of the would healing.

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**TWO NOVEL MISSENSE MUTATIONS OF PERIPHERIN/RDS GENE IN AUTOSOMAL DOMINANT RETINITIS PIGMENTOSA (ADRP), IN PEDIGREES FROM FRANCE**

**SOUIED E[1,2], GERBER S[2], ROZET JM[2], CAMUZAT A[2], DUFIER JL[3], SOUBRANE G[1], COSCAS G[1], MUNNICH A[2] and KAPLAN J[2]**
[1] *Clinique Ophtalmologique Universitaire de CRETEIL (FRANCE)*
[2] *Service de génétique et Unité de Recherches sur les Handicaps Génétiques de l'enfant INSERM U393, Hôpital Enfants-Malades, PARIS (FRANCE)*
[3] *Service d'Ophtalmologie, Hôpital Laënnec PARIS (FRANCE)*

*Purpose .* The peripherin/RDS gene is localized on chromosome 6p12 and the normal product of this gene is a transmembrane glycoprotein that is thought to play a structural role in the photoreceptor outer segments. Hitherto, no peripherin/RDS gene analysis in French ADRP families was published, and this study was undertaken to estimate the part of responsibility of this locus in our pedigrees.
*Methods.* We analyzed 58 probands belonging to unrelated ADRP French families. Our strategy was to analyze the coding sequence of the peripherin/RDS gene using a combination of single-strand conformation polymorphism and direct sequencing analysis.
*Results.* The sequence analyses revealed two previously unreported missense mutations: Cys165Tyr and Phe211Leu in exons 1 and 2 respectively. Cosegregation of the base substitution with the disease could not be tested in the families, but several lines of evidence support the idea that these base substitutions are disease-causing mutations.
*Conclusion.* We report here the identification of two novel mutations in the peripherin/RDS gene of two ADRP probands in French families.

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**A POSITIONAL CLONING STRATEGY TO IDENTIFY THE RP2 GENE.**

**Thiselton D.L.[1] Hardcastle A. J.[1] Nayudu M.[1] Hampson R.M.[1,2] Jones S.E.[2] and Bhattacharya S.S.[1]**
[1] Dept. of Molecular Genetics, Institute of Opthalmology, University of London (UK)
[2] Rayne Institute, St Thomas' Hospital, London (UK)

*Purpose* The X-linked retinitis pigmentosa 2 (RP2) gene is poorly localised (Xp11.3-11.22) with no biochemical clues as to its nature or function. Our aim is to refine the interval containing this gene and create a physical contig of the relevant region, as a resource from which to isolate novel retinal genes as potential candidates for the disorder.
*Methods* Haplotype analysis of xlRP families has been performed to define the disease as RP2 or RP3 (Xp21.1) and to further refine the RP2 critical interval. Contig construction has been achieved using key markers, and novel YAC end sequences in a chromosome walking strategy. A candidate gene has been screened for mutations by direct sequencing in affected males. cDNA selection is being applied to several YACs in the defined genomic interval.
*Results* The RP2 critical region has been reduced by approximately 4 cM with new flanking markers MAOA and DXS6941, thereby excluding several candidate genes. We have established a YAC contig extending from DXS1264 to DXS1126, spanning approximately 3 Mb. This contig is currently being extended distally towards MAOA, where a 1Mb contig has also been constructed. TIMP1 was sequenced in affected males from 6 RP2 families and no mutations were found.
*Conclusions* The genetic refinement of the RP2 critical interval in conjunction with physical mapping data and direct sequencing has enabled us to exclude several candidate genes. The YAC contig in this region provides a means to anchor and order microsatellites and STSs as they are generated. It is also an essential tool from which to isolate novel retinal sequences as candidates for the several ocular diseases mapping to Xp11.23.